Patient ID	Patient Name		Birth Date	Sex	Age
SA00175011	TESTINGRNV, ABNORMAL		2004-03-29	F	20
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
SA00175011	SA00175011	CLIENT,CLIENT			
Account Information		**Collected**			
C7028846 DLMP Rochester		28 Mar 2025 00:00			

MYOD1 Mutation Analysis, Tumor

Result

MCR

Provided diagnosis: soft tissue rhabdomyosarcoma

The following CLINICALLY RELEVANT VARIANT was detected:
Gene: MYOD1
DNA Change: c.365T>G (Exon 1)
Amino Acid Change: p.L122R (Leu122Arg)
Variant Allele Frequency: 87.3%

No other reportable sequence variants were detected within the analyzed regions of the tested gene(s) listed in the method description.

Interpretation

MCR

1) MYOD1 c.365T>G (p.L122R) (Exon 1)

MYOD1 encodes Myoblast determination protein 1, MyoD1, a transcription factor involved in the control of muscle cell differentiation [PMID:16099183, PMID:2155707, PMID:8269513, PMID:8675005]. Although MYOD1 has been suggested to play a tumor suppressor role in several tumor types, a recurrent neomorphic MYOD1 mutation (L122R) has been reported in rhabdomyosarcoma, indicating that the role of MYOD1 in cancer may be context dependent [PMID:24092238, PMID:14767572, PMID:19750229, PMID:14760078, PMID:27284360, PMID:1329087, PMID:24793135, PMID:24272621].

MYOD1-mutant spindle cell/sclerosing rhabdomyosarcoma is a distinct entity in the 2020 WHO classification of soft tissue and bone tumors [WHO Classification of Tumours Editorial Board. Soft Tissue and Bone Tumours. Lyon (France): International Agency for Research on Cancer, 5th ed., vol. 3, 2020]. Mutations in MYOD1, specifically the recurrent alteration L122R, have been associated with a poor prognosis in spindle cell/sclerosing rhabdomyosarcoma [PMID: 30181563, PMID 33913590].

Additional studies are needed in order to assess the clinical implications of MYOD1 testing in other neoplasms.

There are currently no known clinically approved therapies that specifically target MYOD1 mutations.

Additional Information

MCR

CLINICAL TRIALS

Possible clinical trials of benefit for this patient can be found at the following sites:
1) ClinicalTrials.gov:
www.clinicaltrials.gov/ct2/search/advanced
2) Mayo Clinic: www.mayo.edu/research/clinical-trials/ 3) National Cancer Institute: www.cancer.gov/clinicaltrials/search

REFERENCE TRANSCRIPT

Sequence variant nomenclature is based on the following RefSeq accession number (build GRCh37 (hg19)): MYOD1 NM_002478.

Specimen

MCR

Tissue, Tumor

Tissue ID

MCR

12345

Method

MCR

Microscopic examination is performed by a pathologist to identify areas of tumor for enrichment by macrodissection. DNA is extracted from FFPE or cytology slides, and next generation sequencing is performed to evaluate the presence of a mutation in all coding regions and exon/intron boundaries of the MYOD1 gene

Variant nomenclature is based on build GRCh37 (hg19). For details about gene transcripts (RefSeq accession numbers), specific targeted regions of each gene, and additional information on this test, see www.mayocliniclabs.com (Test ID MYODT).

Disclaimer

1 MCR

This test cannot differentiate between somatic and germline

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292

Patient ID	Patient Name		Birth Date	Sex	Age
SA00175011	TESTINGRNV, ABNORMAL		2004-03-29	F	20
Order Number	Client Order Number	Ordering Physician	Report Notes		
SA00175011	SA00175011	CLIENT,CLIENT			
Account Information		Collected			
C7028846 DLMP Rochester		28 Mar 2025 00:00			

alterations. Additional testing may be necessary to clarify the significance of results if there is a potential hereditary risk.

DNA variants of uncertain significance may be identified.

A negative result does not rule out the presence of a variant that may be present but below the limits of detection of this assay. The analytical sensitivity of this assay for sequence reportable alterations is 5% mutant allele frequency with a minimum coverage of 500X in a sample with at least 20% tumor content.

Point mutations and small insertion/deletion mutations will be detected in the MYOD1 gene only. This test may detect single exon deletions but does not detect multi-exon deletions, duplications, larger-scale genomic copy number variants, copy neutral loss of heterozygosity (LOH), or epigenetic modifications such as promoter methylation. DELINS of 1,000 bp or less are detectable with at least 50 or more supporting reads.

Variant allele frequency (VAF) is the percentage of sequencing reads supporting a specific variant divided by the total sequencing reads at that position. In somatic testing, VAF should be interpreted in the context of several factors including, but not limited to: tumor purity/heterogeneity/copy number status (ploidy, gains/losses, loss of heterozygosity) and sequencing artifact/misalignment [PMID: 27144058, PMID: 29769535].

Rare polymorphisms may be present that could lead to false-negative or false-positive results.

The presence or absence of a variant may not be predictive of response to therapy in all patients.

Test results should be interpreted in the context of clinical, tumor sampling, histopathological, and other laboratory data. If results obtained do not match other clinical or laboratory findings, contact the laboratory for discussion. Misinterpretation of results may occur if the information provided is inaccurate and/or incomplete.

Reliable results are dependent on adequate specimen collection and processing. This test has been validated on cytology slides and formalin-fixed, paraffin-embedded tissues; other types of fixatives are discouraged. Improper treatment of tissues, such as decalcification, may cause PCR failure.

Released By MCR

Gang Zheng, M.D., Ph.D.

Received: 29 Mar 2025 16:34 **Reported:** 04 Apr 2025 18:04

Laboratory Notes

1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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